



MTTP gene

microsomal triglyceride transfer protein

Normal Function

The *MTTP* gene provides instructions for making a protein called microsomal triglyceride transfer protein. The microsomal triglyceride transfer protein helps produce beta-lipoproteins, which are molecules that consist of proteins combined with cholesterol and particular types of fats called phospholipids and triglycerides. These beta-lipoproteins contain the protein apolipoprotein B, which is critical for the creation of molecules called chylomicrons. Chylomicrons are formed when dietary fats and cholesterol are absorbed from the intestines. Chylomicrons are also necessary for the absorption of certain fat-soluble vitamins, especially vitamins E, A, and sometimes K.

Other beta-lipoproteins containing apolipoprotein B, particularly low-density lipoproteins (LDL) and very low-density lipoproteins (VLDL), are created by microsomal triglyceride transfer protein in the liver. These lipoproteins transport fats, cholesterol, and fat-soluble vitamins throughout the body in the bloodstream. Sufficient levels of fats, cholesterol, and vitamins are necessary for normal growth, development, and maintenance of the body's cells and tissues, particularly nerve cells and tissues in the eye.

Health Conditions Related to Genetic Changes

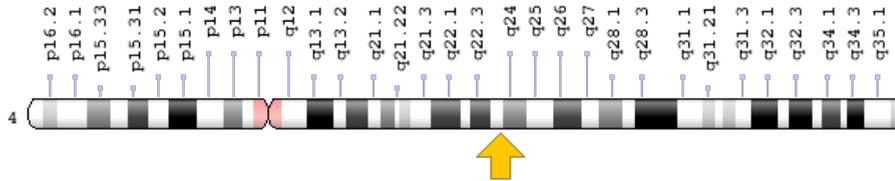
abetalipoproteinemia

More than 30 mutations that cause abetalipoproteinemia have been identified in the *MTTP* gene. One particular mutation is more common in people of Ashkenazi (eastern and central European) Jewish descent; this mutation replaces the protein building block (amino acid) glycine with a stop signal at position 865 (written as Gly865X or G865X). As a result of this amino acid change, an abnormally small, nonfunctional version of the protein is made. Similarly, many other mutations that cause abetalipoproteinemia produce abnormally short microsomal triglyceride transfer proteins. A shortened protein is unable to produce beta-lipoproteins, resulting in a severe deficiency of chylomicrons, low-density lipoproteins, and very low-density lipoproteins. The absence of these lipoproteins prevents dietary fats and fat-soluble vitamins from being absorbed and carried to the rest of the body, leading to the nutritional and neurological problems seen in people with abetalipoproteinemia.

Chromosomal Location

Cytogenetic Location: 4q23, which is the long (q) arm of chromosome 4 at position 23

Molecular Location: base pairs 99,564,078 to 99,623,997 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ABL
- microsomal TG transfer protein
- microsomal triglyceride transfer protein (large polypeptide, 88kD)
- microsomal triglyceride transfer protein large subunit
- MTP
- MTP triglyceride carrier
- MTP_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Lipoproteins Transport Cholesterol and Triacylglycerols Throughout the Organism
<https://www.ncbi.nlm.nih.gov/books/NBK22336/#A3634>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MTTP%5BTIAB%5D%29+OR+%28microsomal+triglyceride+transfer+protein%5BTIAB%5D%29%29+A+ND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MICROSOMAL TRIGLYCERIDE TRANSFER PROTEIN
<http://omim.org/entry/157147>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MTTP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MTTP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7467
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4547>
- UniProt
<http://www.uniprot.org/uniprot/P55157>

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